

Variant: NM_000314.6(PTEN):c.75G>A (p.Leu25=)

CA000197 [↗](#)

184505 (ClinVar) [↗](#)

Gene: PTEN ([HGNC:5728](#))

Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: e5e35031-cdfe-44ec-ad5e-0f3892c0a7b1

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HGVS expressions

NM_000314.6:c.75G>A

NM_000314.6(PTEN):c.75G>A (p.Leu25=)

NC_000010.11:g.87864544G>A

CM000672.2:g.87864544G>A

NC_000010.10:g.89624301G>A

CM000672.1:g.89624301G>A

NC_000010.9:g.89614281G>A

NG_007466.2:g.6106G>A

NG_033079.1:g.3894C>T

ENST00000686459.1:c.75G>A

ENST00000688158.1:c.75G>A

ENST00000688308.1:c.75G>A

ENST00000693560.1:c.594G>A

ENST00000371953.8:c.75G>A

ENST00000371953.7:c.75G>A

ENST00000462694.1:n.77G>A

ENST00000487939.1:n.96G>A

ENST00000610634.1:c.-28G>A

ENST00000618586.1:n.44G>A

NM_000314.5:c.75G>A

NM_001304717.2:c.594G>A

NM_001304718.1:c.-631G>A

NM_000314.7:c.75G>A

NM_001304717.5:c.594G>A

NM_001304718.2:c.-631G>A

NM_000314.8:c.75G>A

NM_000314.8(PTEN):c.75G>A (p.Leu25=)

Likely Benign

Met criteria codes 3

BS3

PM2_Supporting

BP7

Not Met criteria codes 23

BS2

PVS1

BS4

BS1

BP2

BP3

BP4

BP1

BP5

PS2

PS4

PS3

PS1

BA1

PP4

PP1

PP3

Expert Panel

PTEN VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen PTEN Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines for PTEN Version 3.0.0

Evidence submitted by expert panel

PTEN VCEP

NM_000314.8(PTEN):c.75G>A (p.Leu25=) meets criteria to be classified as likely benign for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.0.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). **BS3: Synonymous variant with RNA, mini-gene, or other splicing assay demonstrating no splicing impact. (internal laboratory contributor)** **BP7: Variant is synonymous and no splicing impact is predicted. PM2_P: Absent in gnomAD. Using the Bayesian point system (PMID: 29300386) for this variant with conflicting evidence: 1 benign strong, 1 benign supporting and 1 pathogenic supporting codes get -4 + (-1) + 1 points; total is - 4 (likely benign).**

Met criteria codes

BS3			Synonymous variant with RNA, mini-gene, or other splicing assay demonstrating no splicing impact. (internal laboratory contributor)
PM2_Supporting			Absent in gnomAD.
BP7			Variant is synonymous and no splicing impact is predicted.

Not Met criteria codes

BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2			1 internal lab observation in cis with D24N (LPATH per this lab), need 3 observations in cis or phase unknown with different variants to apply BP2.
BP3			

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP4			In silico predictors unable to provide prediction for nearby SDS.
BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			1 internal case from CCF PTEN study with CC Score of 22. OFC: unknown; Papillary Thyroid Carcinoma dx age 15; Fibrocystic Breast Disease; Left breast cancer, dx 37- ductal carcinoma + left DCIS (cribriform), mastectomy; Fibroadenoma of Breast; Uterine Fibroids; Fibroma; Oral Mucosa Papilloma. Doesn't meet criteria for PS4, PS4_moderate, or PS4_supporting.
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			See PS4.
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 		
    		
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